

FIGURE 1

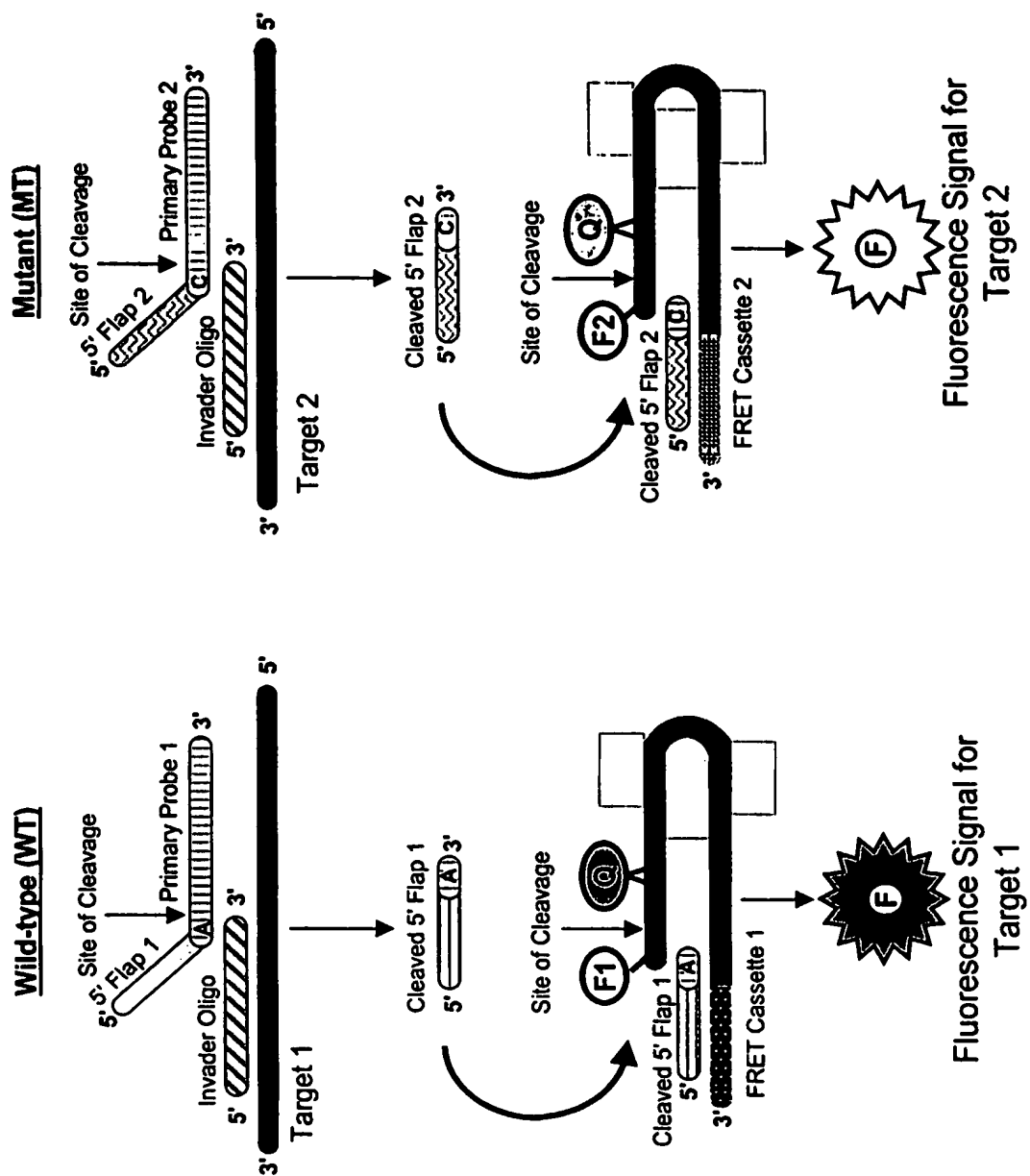


FIGURE 2

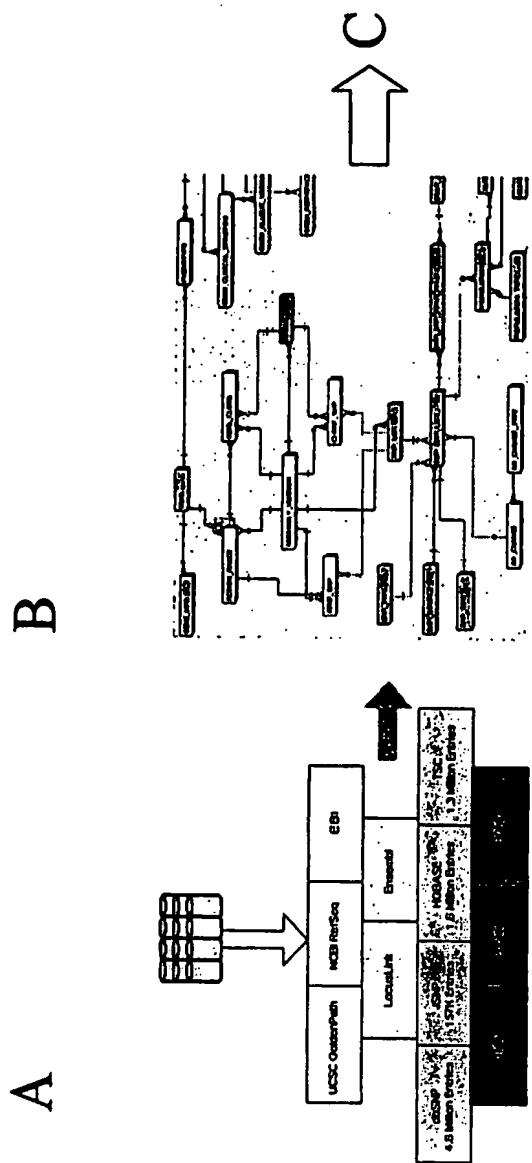
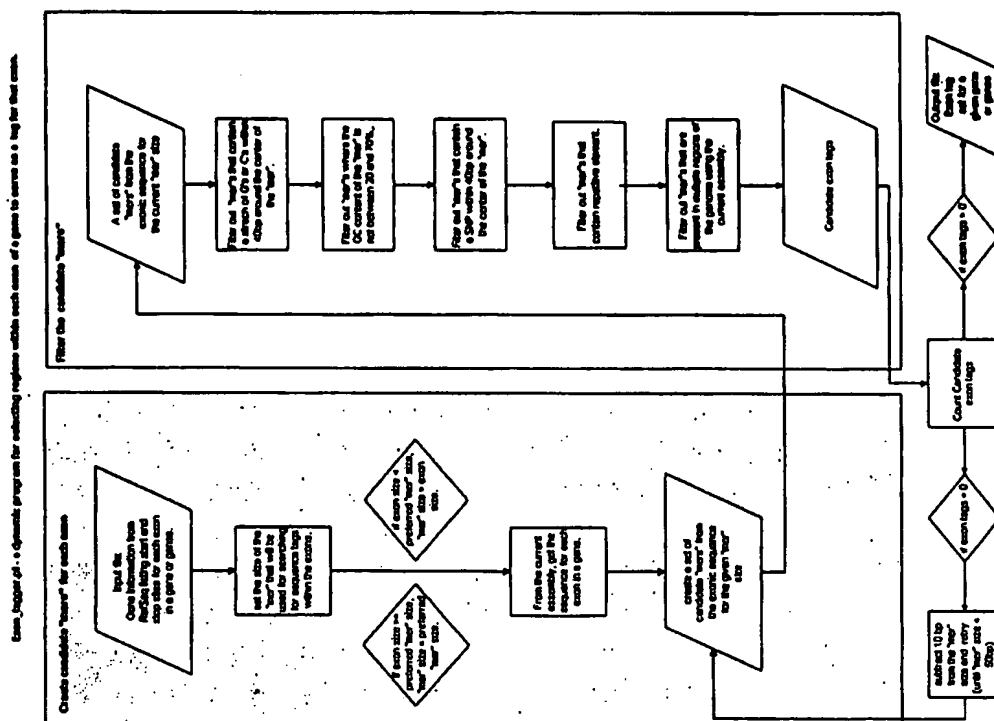


FIGURE 2 (continued)



U

↑
B

Figure 3

Chrom	Gene	%GC	Probe Oligo Sequence	SEQ ID NO	Invader Oligo Sequence	SEQ ID NO
1	ACTA1	N/A	ACGGACCGGGAGGAGCCCTGTGACAT	1	CCATCCAGGGAAGAGTGGCCTGTTT	100
1	ACTA1	N/A	ACGGACCGGGAGGAGCCCTGTGACATT	2	CCATCCAGGGAAGAGTGGCCTGTTT	101
1	ACTA1	N/A	ACGGACCGGGAGGAGCCCTGTGACATT	3	CCATCCAGGGAAGAGTGGCCTGTTT	102
1	ACTA1	N/A	ACGGACCGGGAGGAGCCCTGTGACATT	4	CCATCCAGGGAAGAGTGGCCTGTTT	103
1	ACTA1	N/A	ACGGACCGGGAGGAGCCCTGTGACATT	5	CGTTGGCAGCCCATCCAGGGAAGAT	104
1	ACTA1	54%	ACGGACCGGGAGGAGCCCTGTGACATT	6	GGCGGGCTGAGCTCCAGCCAT	105
1	ACTA1	58%	ACGGACCGGGAGGAGGAGGTGGAGTGTG	7	TTCTGCTCTCTGAGAAAGTCGCGTGCC	106
1	ACTA1	63%	ACGGACCGGGAGGAGGAGGTGGAGTGTG	8	GGCGCTGGACCTGGCGGGCT	107
1	ACTA1	62%	ACGGACCGGGAGGAGGAGGTGGAGTGTG	9	GGCGGCTTTCACGAGGCGCGGAT	108
1	HIST2H2BE	58%	ACGGACCGGGAGGAGGAGGTGGAGTGTG	10	GGCCCATCGCACGGCAGCAACTT	109
1	HIST2H2BE	56%	ACGGACCGGGAGGAGGAGGTGGAGTGTG	11	ACAAATTCAGCCCGGGTTCGCGAAACAA	110
1	SV2A	59%	ACGGACCGGGAGGAGGAGGTGGAGTGTG	12	CAGGGCCAGAGAGCTGCCAAGGG	111
1	CRA	59%	ACGGACCGGGAGGAGGAGGTGGAGTGTG	13	TCAGCTCTGCTCTTGGCGGGACAGTCC	112
21	DSCR8	60%	CGCGCCGAGGCAAGAAATTCATGTCTCAG	14	CACATGACGCTCCACCTCCAGTTT	113
21	DSCR6	52	CGCGCCGAGGCTCGACTCACGGCA	15	CCAGGCTGGAATGCAATGGTGCGATT	114
21	DSCR8	58	CGCGCCGAGGCTCGACTCACGGCA	16	ACTGGAGGTGGAGGCTGCAGTGATT	115
21	DSCR6	56	CGCGCCGAGGCTCGACTCACGGCA	17	GGCCTCCAGTCTGTGTCAGCTCAGCATT	116
21	DSCR1	58%	CGCGCCGAGGCTCGACTCACGGCA	18	GGTCTCTGTGTGCAAGGGCAGGTGATT	117
21	AML1	56%	CGCGCCGAGGCTCGACTCACGGCA	19	GCCTCAGTGGAGACAAGTGGGAAACATGGTT	119
21	AML1	54%	CGCGCCGAGGCTCGACTCACGGCA	20	TGGACGTGCCAGCGGCATGACAT	120
21	AML1	57%	CGCGCCGAGGCTCGACTCACGGCA	21	CTTACAAACCCACGCAAGTCGCCAT	121
21	DSCR9	57%	CGCGCCGAGGCTCGACTCACGGCA	22	CTCCACCCGTTCCATCCACGGCT	122
21	DSCR9	58%	CGCGCCGAGGCTCGACTCACGGCA	23	GCACCTCCACCCGTTCCATCCCT	123
X	L1CAM	58%	CGCGCCGAGGCTCGACTCACGGCA	24	CGGATTCAGCGTGGCGCCCTGATT	124
X	L1CAM	58%	CGCGCCGAGGCTCGACTCACGGCA	25	CGGATTCAGCGTGGCGCCCTGATT	125
X	L1CAM	58%	CGCGCCGAGGCTCGACTCACGGCA	26	CGGATTCAGCGTGGCGCCCTGATT	126
X	L1CAM	58%	CGCGCCGAGGCTCGACTCACGGCA	27	CGGATTCAGCGTGGCGCCCTGATT	127
X	L1CAM	58%	CGCGCCGAGGCTCGACTCACGGCA	28	CGGATTCAGCGTGGCGCCCTGATT	128
X	L1CAM	58%	CGCGCCGAGGCTCGACTCACGGCA	29	GATTCAGCGTGGCGCCCTGATT	129
X	L1CAM	58%	CGCGCCGAGGCTCGACTCACGGCA	30	CGGATTCAGCGTGGCGCCCTGATT	130
X	PDCD8	52%	CGCGCCGAGGCTCGACTCACGGCA	31	CAGATTTTGGTGGCTTCCGGGTAATGCAGAT	131
X	PPEF1	53%	CGCGCCGAGGCTCGACTCACGGCA	32	TGCTGGCCTTCAGTCGAGCTTTGTAACCTT	132
Y	SRY	56%	CGCGCCGAGGCTCGACTCACGGCA	33	GCCTCCCGCAGATCCCGCTTCGGTAT	133
Y	EIF1A1	54%	CGCGCCGAGGCTCGACTCACGGCA	34	GACCTCTCCGACTCCTTTCTGGCGGTACTAT	134
18	GATA6	57%	CGCGCCGAGGCTCGACTCACGGCA	35	CACAAGCATGCACAGGGTTCACCCCTT	135
18	SERPINB2	57%	CGCGCCGAGGCTCGACTCACGGCA	36	CTGCCACAACTGGGGCTCCATGTT	136
13	DLEU1	56%	CGCGCCGAGGCTCGACTCACGGCA	37	AGGAGAGCGGTGCACACGATGAC	137
13	ABCC4	65%	CGCGCCGAGGCTCGACTCACGGCA	38	CGGCTGGCTGTGATCAGACTGCCGT	138
13	POU4F1	65%	CGCGCCGAGGCTCGACTCACGGCA	39	CGTGGGCTCACTCAGCCAGAGCAT	139
13	POU4F1	63%	CGCGCCGAGGCTCGACTCACGGCA	40	CTGAGACACAAGTACCCGCTGCTGCAT	140
13	POU4F1	54%	CGCGCCGAGGCTCGACTCACGGCA	41	CCTCGTCCGAGAAAGATCGCCGCCATCT	141
13	POU4F1	54%	CGCGCCGAGGCTCGACTCACGGCA	42	CCACTCACTTCCCGGGATTGGAGAGCAT	141

Aneuploidy Probe and Invader Designs

13	PCDH9	CGCGCCGAGGCACTGTCACATGCAGC	57%	43	ACTACCCACCGGCTCATGGTCTCCTAGACT	142
13	PCDH9	CGCGCCGAGGCTGCACCTCTGAGGCA	54%	44	GGCCTTTGCTTGAAGCCTCTCTGGGAAT	143
13	PCDH9	CGCGCCGAGGCACTCTCTGCTCGA	54%	45	ACAGTGCTGGTAGTGACGGCAGCTT	144
13	PCDH9	CGCGCCGAGGCTCGTGGTAGAGAA	51%	46	GTGCCCTCTCAGCCATTCTCTGGCC	145
18	FLJ23403	CGCGCCGAGGCTGGATCTGTGTGG	52%	47	TGGACGGACACAAGCTTTGAGCCTGTCCAT	146
18	KIA0222	CGCGCCGAGGCTGGAGACCAGATCA	56%	48	CGAAGAGGTGACTTCGACCGAGCTCTCCAT	147
21	CLDN17	CGCGCCGAGGCATGCACAGATGCC	56%	49	CGTAGAGCCTGTGCACCTGACCTGCTTT	148
21	MGC33295	CGCGCCGAGGCATGCTTCGGAGACTC	56%	50	CTCCAGTCAGGCTCCACACGCT	149
21	PCP4	CGCGCCGAGGCTGGTGCATCCATGTC	52%	51	GGCCACCGCTGCACGTTCTGTCTT	150
X	PFKFB1	CGCGCCGAGGCTGCTCCCTGAAGGT	56%	52	TGGCCAATTCATTAGTCCACGGGCATT	151
X	PFKFB1	CGCGCCGAGGCTGCTCCCTGAAGGT	56%	53	TGGCCAATTCATTAGTCCACGGGCATT	152
X	PFKFB1	CGCGCCGAGGCTGCTCCCTGAAGGT	56%	54	GTGACTGGTCCACACCTTCAGGGAGCTT	153
X	ZNF157	CGCGCCGAGGCTGCTCCCTGAAGGT	60%	55	GTTACCTCGGCTCAGCCCAT	154
X	FLJ22843	CGCGCCGAGGCTTCCGTGCTGGA	62%	56	CTGCTCTGCTCCGGGCAACT	155
X	DUSP21	CGCGCCGAGGCTTCAACCGCAAC	59%	57	GAGCGGAACGGCTCACTCCAGCT	156
X	DUSP21	CGCGCCGAGGCTGCAAGTGCAGCA	59%	58	GAGCGGAACGGCTCACTCCAGCT	157
X	MGC33889	CGCGCCGAGGCTGCTAGCTCCCA	57%	59	GGTGACTCTCTTCTGCCACAGTGGCT	158
Y	PRKY	CGCGCCGAGGCTGCTAGCTCCCA	48%	60	TGCTCCCACTTCTTCCCAAGAGAACTCTCT	159
Y	PRKY	CGCGCCGAGGCTGCTAGCTCCCA	55%	61	GCACCCAGTAGGGTGGAGTATACCTTT	160
Y	PRKY	CGCGCCGAGGCTGCTAGCTCCCA	53%	62	GTCCCTCGGCTCTTAATCTCTCAAACTCAGC	161
Y	TMSB4Y	CGCGCCGAGGCTGCTAGCTCCCA	59%	63	TCGGACTCCCTAGCACACATGCTCTT	162
Y	TMSB4Y	CGCGCCGAGGCTGCTAGCTCCCA	59%	64	GCACAGTCCAGTACAGAGACCCGGAT	163
21	NRIP1	CGCGCCGAGGCTGCTAGCTCCCA	54%	65	GCTCTCTGTGCAGAGAGCGGCACT	164
21	NRIP1	CGCGCCGAGGCTGCTAGCTCCCA	54%	66	GCTCTCTGTGCAGAGAGCGGCACT	165
21	HLCS	CGCGCCGAGGCTGCTAGCTCCCA	60%	67	TGACCCGAGGCAACTCTCTCT	166
21	HLCS	CGCGCCGAGGCTGCTAGCTCCCA	57%	68	GCCACGATTGACTTCTACGAGTACTGATGAT	167
21	HLCS	CGCGCCGAGGCTGCTAGCTCCCA	58%	69	CCGTTCCATCCAGGCGAGCAGTT	168
21	HLCS	CGCGCCGAGGCTGCTAGCTCCCA	55%	70	AAGTGGCTTGACGCCATCTCTGGATT	169
21	HLCS	CGCGCCGAGGCTGCTAGCTCCCA	56%	71	GTGGAAGGTTTGTCTTGTGCCAGGCT	170
21	HLCS	CGCGCCGAGGCTGCTAGCTCCCA	56%	72	GTGGAAGGTTTGTCTTGTGCCAGGCT	171
21	HLCS	CGCGCCGAGGCTGCTAGCTCCCA	59%	73	TGCCGAGCTCACCGTGTCTCAT	172
18	CN2	CGCGCCGAGGCTGCTAGCTCCCA	54%	74	GGAGAAGAGAGCGGAAATCAGGAGGATGATGAAT	173
21	HLCS	CGCGCCGAGGCTGCTAGCTCCCA	64%	75	ACCAGAGCGAGGTGAAGCTCAGCGTT	174
21	HLCS	CGCGCCGAGGCTGCTAGCTCCCA	64%	76	TCCTGGTAGCTGCAGCCACTGCTCAAT	175
21	HLCS	CGCGCCGAGGCTGCTAGCTCCCA	58%	77	CGCTGCTTGAAGCGTTTGCATGGTCT	176
21	HLCS	CGCGCCGAGGCTGCTAGCTCCCA	57%	78	TCCATTTGCCAGGCTTGTCTCATCT	177
21	HLCS	CGCGCCGAGGCTGCTAGCTCCCA	57%	79	TCCATTTGCCAGGCTTGTCTCATCT	178
21	HLCS	CGCGCCGAGGCTGCTAGCTCCCA	57%	80	ACCACCTCATCAGCGAGAACTTCCCGCC	179
21	HLCS	CGCGCCGAGGCTGCTAGCTCCCA	63%	81	GCCTCTGCACACTCAGCAGGATGCT	180
X	MTMR8	CGCGCCGAGGCTGCTAGCTCCCA	59%	82	CCATCTGAGTCTCCCGCCACT	181
X	MGC23947	CGCGCCGAGGCTGCTAGCTCCCA	53%	83	CTCTTTTCTTCCCGCAGTGTGTGAACACAT	182
X	FLJ21174	CGCGCCGAGGCTGCTAGCTCCCA	56%	84	TGGCTCTCAGCTGGCATTGTGACCTT	183
X	ESX1L	CGCGCCGAGGCTGCTAGCTCCCA	62%	85	CCGACCCGCTTCAAGGATTTCT	184
X	ZNF157	CGCGCCGAGGCTGCTAGCTCCCA	52%	86	CTGGTATCATATGCCCCCAAGGAGCTTGATCT	185

Aneuploidy Probe and Invader Designs

X	PCTK1	57%	CGCGCCGAGGTGAAGCCTGGTCACT	187	CTGCACCTCATCCGAGGACGTGGCC	186
X	MGC33889	62%	CGCGCCGAGGTGCGTAGCTCCAG	188	GGTGACTCTCCTTCTGCCACAGTGGCCC	187
21	DSCR8	57%	CGCGCCGAGGGAGCAGTCTGTAACTG	189	CTCCCCACCGTTCCATCCAGGCT	188
21	DSCR9	57%	CGCGCCGAGGAGGCGAGAGTCTG	190	GCACCTCCCCACCGTTCATCCCT	189
21	DSCR9	57%	CGCGCCGAGGCCATCCAGGCGA	191	GTCTGCGCACCTCCCACCGTTT	190
21	DSCR9	57%	CGCGCCGAGGTTCCATCCAGGCG	192	CGCTGCGCACCTCCCCACCGC	191
Y	SRV	55%	CGCGCCGAGGACTGTACGAAAGCCAC	193	GAAGTGCAACTGGACAACAGGTTGTACAGGGATT	192
Y	SRV	55%	CGCGCCGAGGAGGTTGTACAGGGATGA	194	GGTACTCTGCAGCGAAGTGCAACTGGACAAC	193
Y	SRV	56%	CGCGCCGAGGGCAGATCCCGCTTC	195	TGCTGCCGAAGATTGCAGTTTGTCTCCCT	194
Y	SRV	54%	CGCGCCGAGGCTTCGGTACTCTGC	196	GAAGAAITGCAGTTTGTCTCCCGCAGATCCCT	195
21	CLDN17	59%	CGCGCCGAGGTGAAGCAGGTCCAGT	197	CGCCCTGCTTATTGGCATCTGTGGCAC	196
21	DSCR10	56%	CGCGCCGAGGGCATGGCTCTGCAC	198	GCTTGAAGGTGACACACACAGTGACACGGGAT	197
21	MGC33295	57%	CGCGCCGAGGCCAGTCAGGCCTCC	199	GCAAAGTAAGGAGTCGCATCCCTGCCTCT	198
21	STCH	54	CGCGCCGAGGCCACGCTTGGTGATAG	200	GCCTATGGTCTCCACAAGGCTGACGTCTTT	199
21	STCH	53%	CGCGCCGAGGCTTGGATAGCCACTC	201	GAGAGCCCAAGAGCCTCCATCAATCCCTT	200
X	AR	56%	CGCGCCGAGGCCAGTCTCCGGA	202	CGACTCTGGTACGAGCTGCCTCGTT	201
13	CCNA1	55%	CGCGCCGAGGGCGCATGCCCTCC	203	TCCACGACCGAGTCCACCAAGATCGTT	202
13	CCNA1	49%	CGCGCCGAGGCGTAGGAACAGCAGC	204	GTCTGTCTGAGAGGGAAACTGCAGCTT	203
13	ING1	52%	CGCGCCGAGGTACAGGACCAATCGT	205	GCTAGGCCCGCTTTCATCCGCG	204
21	ADAMTS5	58%	CGCGCCGAGGCCAGCAGTGCAACC	206	GAATCCCGAGGACAGACCTACGATGCCACT	205

SEQ ID NO:199
SEQ ID NO:200

FAM FRET cassette
RED FRET cassette

Fam-TCT-228-AGCCGGTTTTCCGGCTGAGACCTCGGCGCG-hex
Red-TCT-228-TCGGCCCTTTTGGCCGAGAGACTCCGGCTCCGT-hex

FIGURE 4

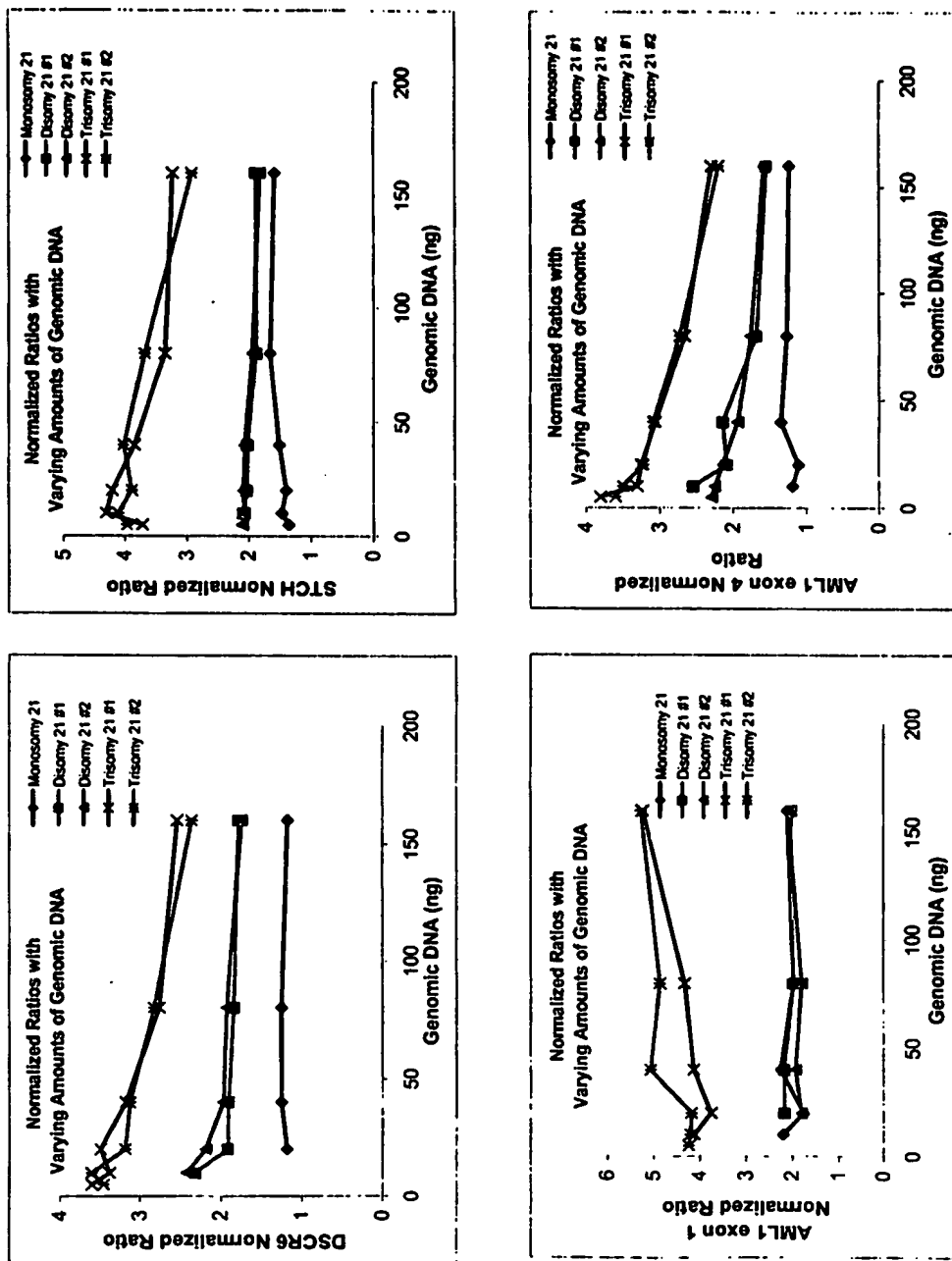


FIGURE 5

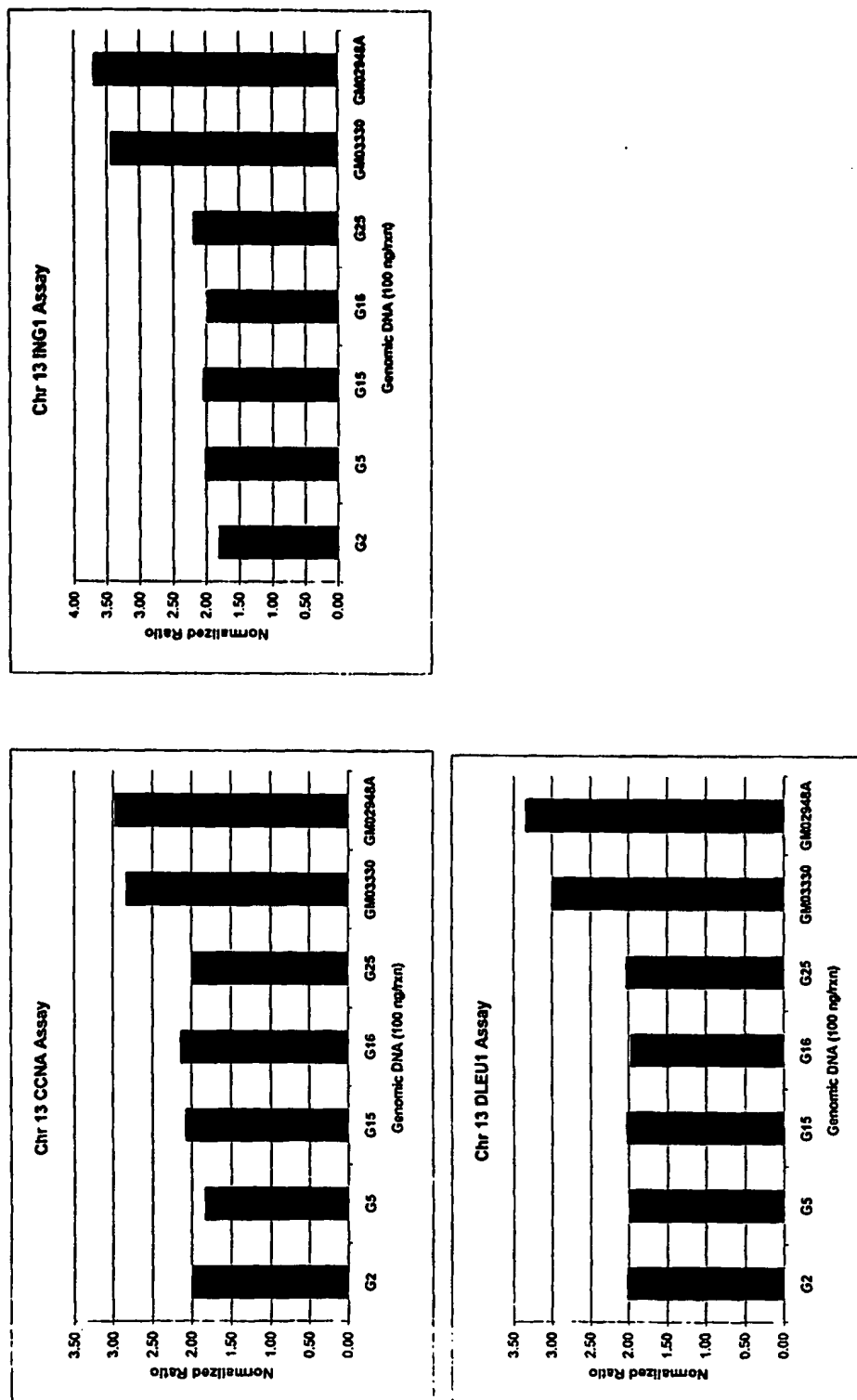


FIGURE 6

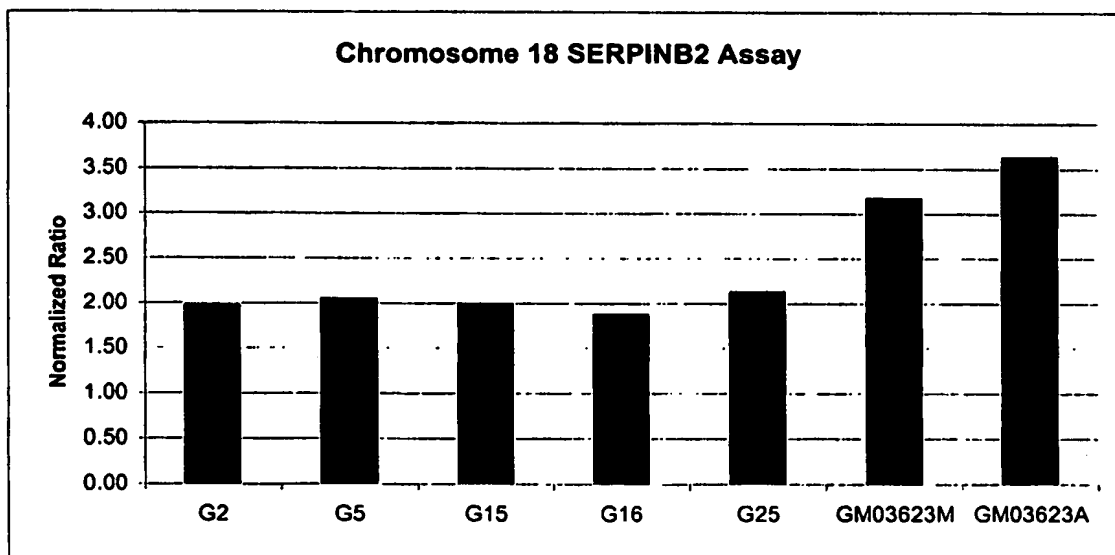
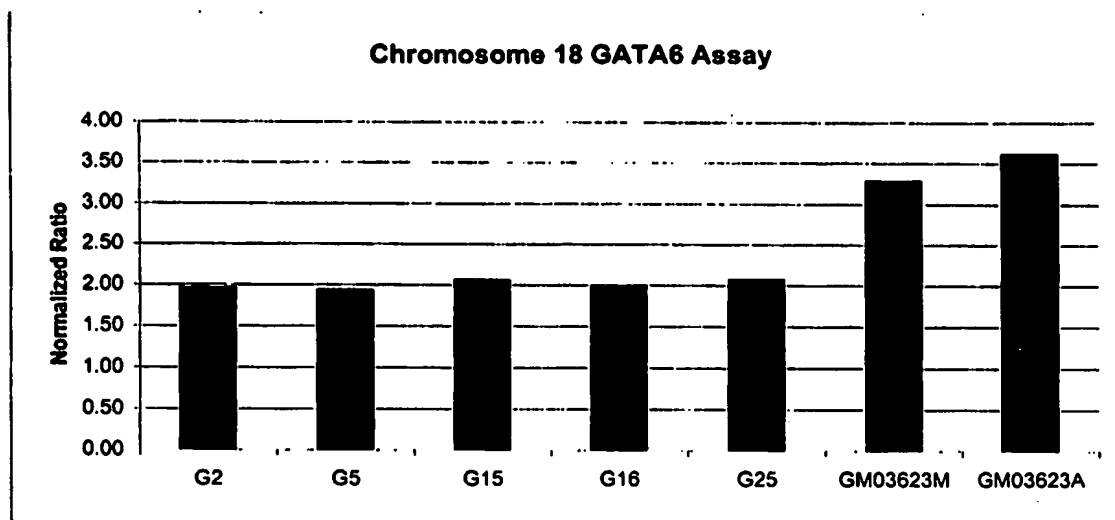


FIGURE 7

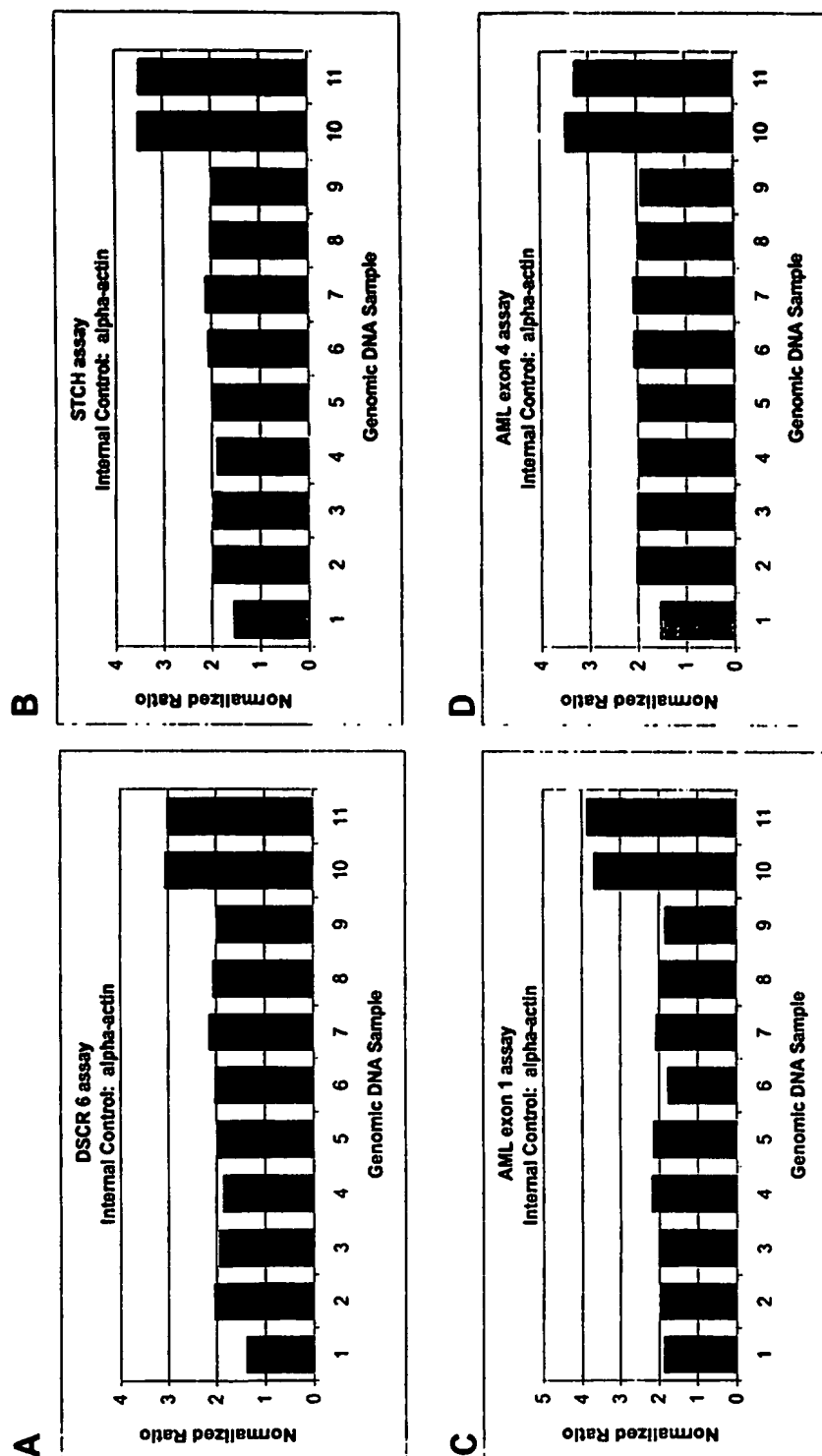


FIGURE 7 (continued)

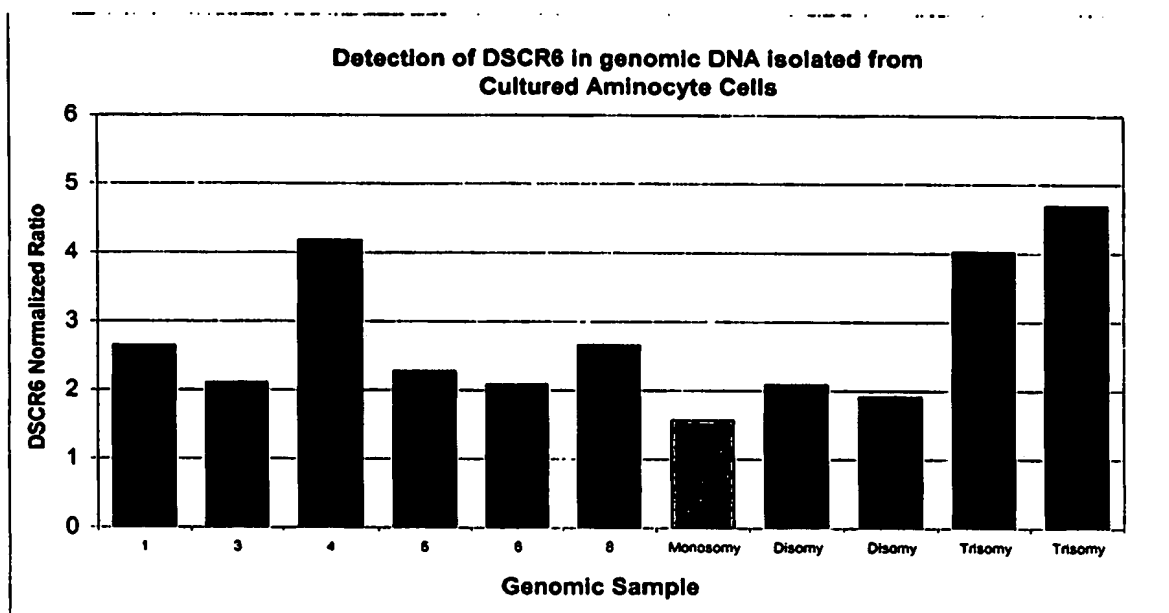
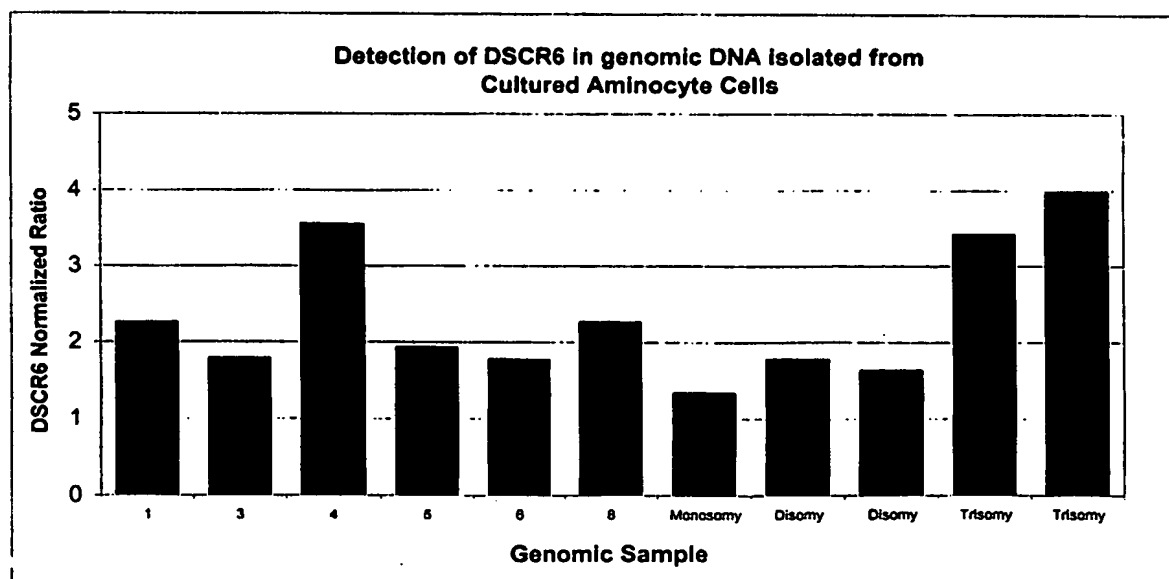
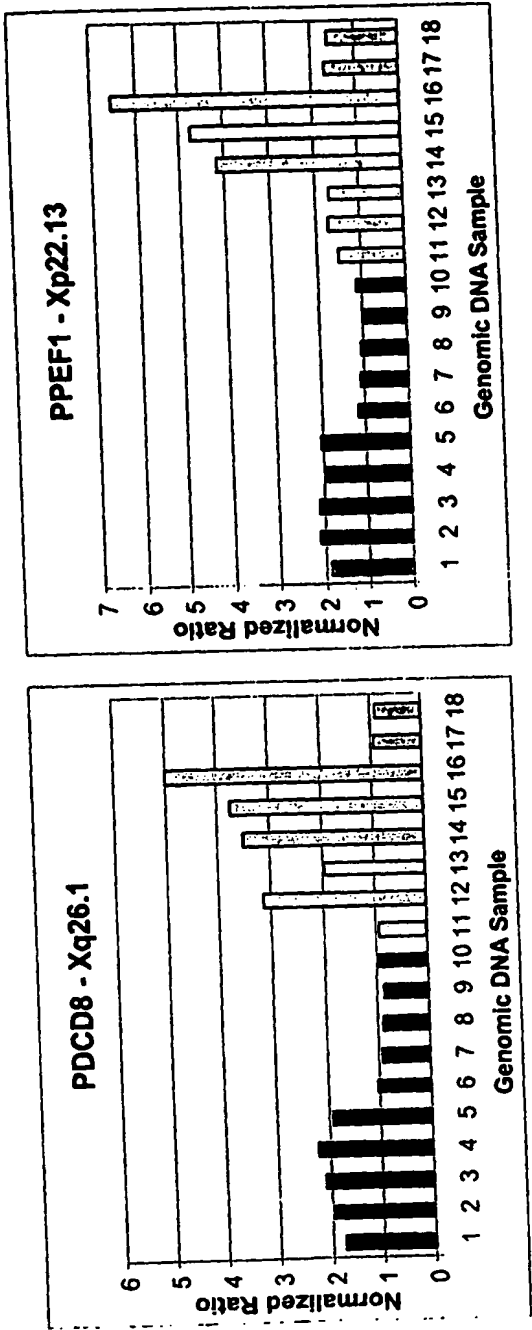
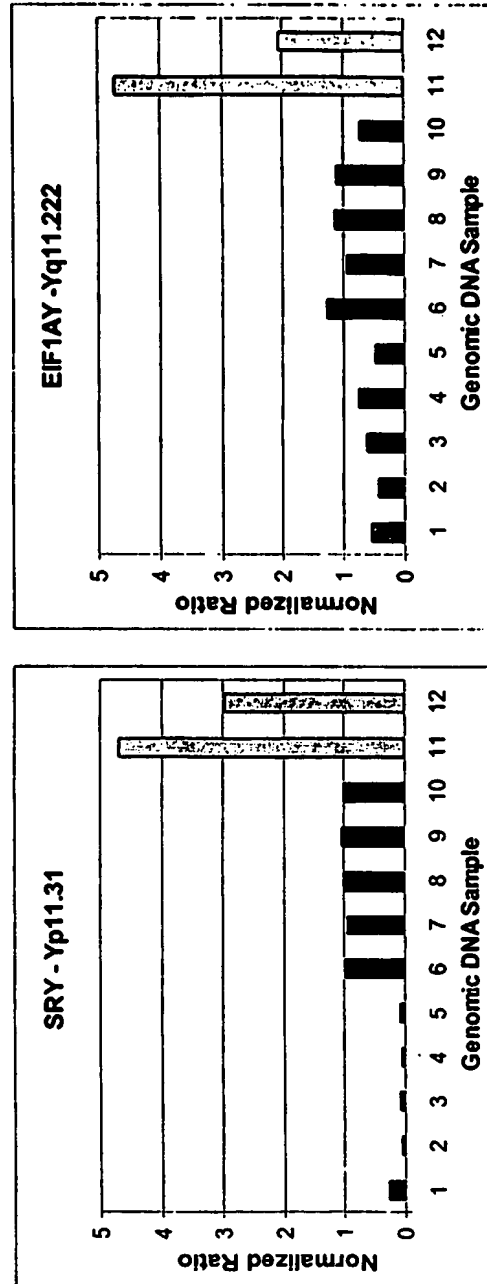
E**F**

FIGURE 8



<u>Sample #</u>	<u>Description</u>
1-5	46, XX
6-10	46, XY
11	45, X
12	30% 45, X / 70% 46, X, iX (qter>cen>Qter)
13	40% 45, X / 60% 46, X, iX (qter>cen>Qter)
14	48, XXX, +18
15	48, XXXX
16	48, XXXX / 49, XXXXX
17-18	47, XYY

FIGURE 9



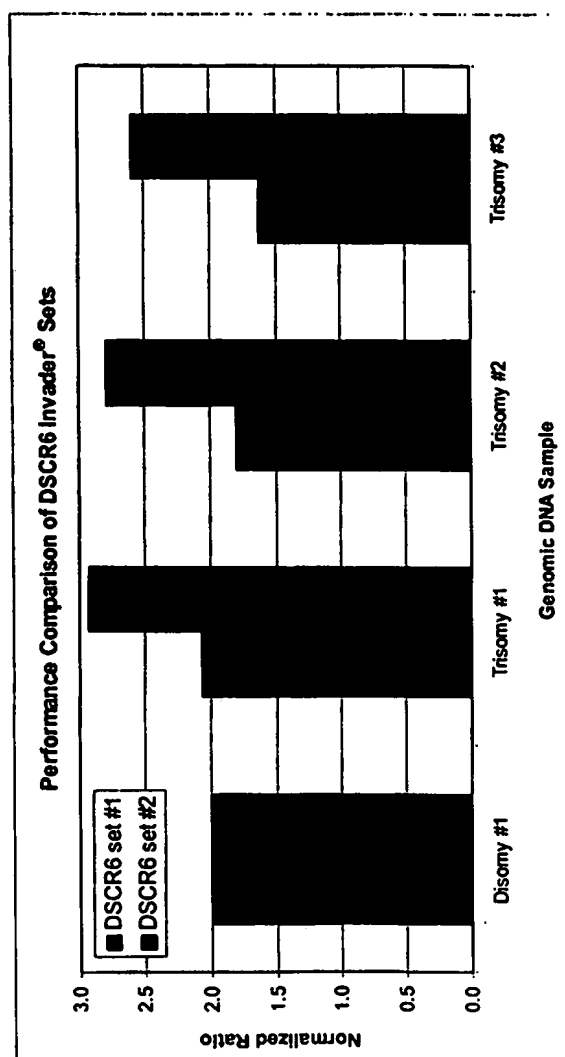
Sample #	Description
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1-5	46, XX
-----	--------

6-10	46, XY
------	--------

11-12	47, XYY
-------	---------

FIGURE 10



Trisomy #1: Coriell AG13429

Trisomy #2: Coriell AG09394

Trisomy #3: Coriell AG10098

-all genomic DNA's were prepped using the Gentra Autopure Prep

FIGURE 11A

STCH Signal (FAM)																												
% Maternal Contamination																												
ng	0	50.82	50.83	50.84	50.85	0	0	1	5	10	20	50	80	90	95	99	100	0	1	5	10	20	50	80	90	95	99	100
ng	0	0	0	0	0	50.82	100	99	95	90	80	50	20	10	5	1	0	50	49.5	47.5	45	40	25	10	5	2.5	0.5	0
Std	205	450	482	421	457	876	1024	1143	1086	1156	1122	959	788	805	749	750	785	741	785	803	809	781	639	561	553	518	497	534
Std	235	448	482	418	468	899	1107	1174	1157	1097	1048	940	802	785	758	725	713	803	782	821	777	722	627	578	541	478	497	509
AV	220	449	482	420	463	888	1066	1159	1127	1127	1085	950	784	785	754	738	739	772	794	812	793	752	633	570	547	497	487	522
SD	21	1	0	2	8	16	59	22	43	42	52	13	25	28	6	18	37	44	2	13	23	42	8	12	8	27	0	18
%	10%	0%	0%	1%	2%	2%	6%	2%	4%	4%	5%	1%	3%	4%	1%	2%	5%	6%	0%	2%	3%	6%	1%	2%	2%	5%	0%	3%
Signal/Bkgd	2.04	2.19	1.91	2.10	4.03	4.84	5.27	5.12	5.12	4.93	4.32	3.56	3.57	3.43	3.35	3.36	3.51	3.81	3.69	3.60	3.42	2.88	2.59	2.48	2.26	2.26	2.37	
Net Signal	229	262	200	243	668	846	939	907	907	865	730	584	565	534	518	519	552	574	592	573	532	413	350	327	277	277	302	

Alpha Actin Signal (Red)																												
% Maternal Contamination																												
ng	0	50.82	50.83	50.84	50.85	0	0	1	5	10	20	50	80	90	95	99	100	0	1	5	10	20	50	80	90	95	99	100
ng	0	0	0	0	0	50.82	100	99	95	90	80	50	20	10	5	1	0	50	49.5	47.5	45	40	25	10	5	2.5	0.5	0
Std	107	309	341	299	322	381	439	436	450	490	528	553	497	562	563	586	594	284	289	323	335	342	342	334	342	345	345	368
Std	114	303	359	298	330	385	438	438	446	472	479	573	497	504	559	559	560	295	300	319	320	327	328	324	338	304	339	357
AV	111	308	350	298	326	383	438	437	448	481	504	563	497	533	561	573	577	290	295	321	328	335	335	329	340	325	342	373
SD	5	4	13	2	6	3	2	1	3	13	35	14	0	41	3	19	24	8	8	3	11	11	10	7	3	29	4	22
%	4%	1%	4%	1%	2%	1%	0%	0%	1%	3%	7%	3%	0%	8%	1%	3%	4%	3%	3%	1%	3%	3%	3%	2%	1%	9%	1%	6%
Signal/Bkgd	2.77	3.17	2.69	2.95	3.47	3.96	3.95	4.05	4.35	4.56	5.10	4.50	4.82	5.08	5.18	5.22	2.62	2.67	2.80	2.96	3.03	3.03	2.98	3.08	2.94	3.10	3.37	
Net Signal	198	240	187	216	273	327	327	338	371	393	453	387	423	451	462	467	179	184	211	217	224	225	219	230	214	232	282	
Ratio	0.56	0.59	0.55	0.54	0.57	1.23	1.30	1.44	1.35	1.23	1.11	0.81	0.73	0.67	0.59	0.56	1.55	1.57	1.41	1.33	1.19	0.92	0.80	0.72	0.65	0.60	0.56	
AV	2.10	1.96	1.91	2.02	4.40	4.64	5.16	4.82	4.39	3.85	2.89	2.62	2.40	2.13	2.01	2.00	5.53	5.59	5.05	4.74	4.26	3.30	2.87	2.56	2.32	2.15	2.07	
Normalized Ratio																												

FIGURE 11B

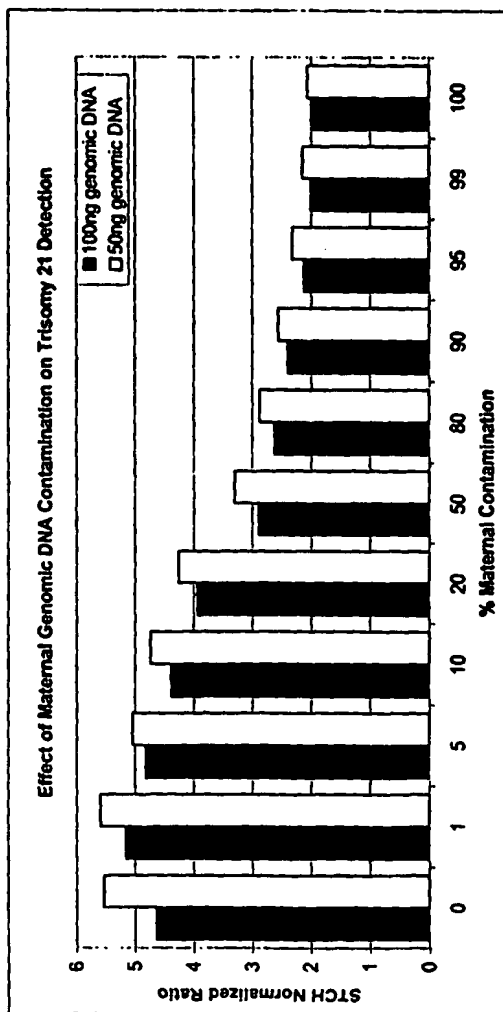
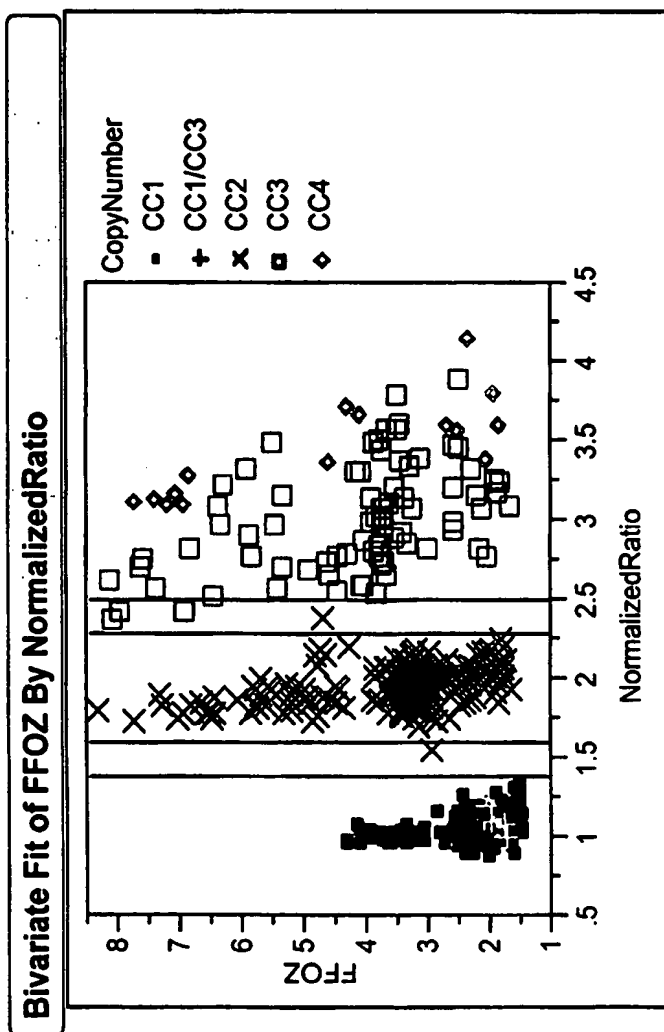


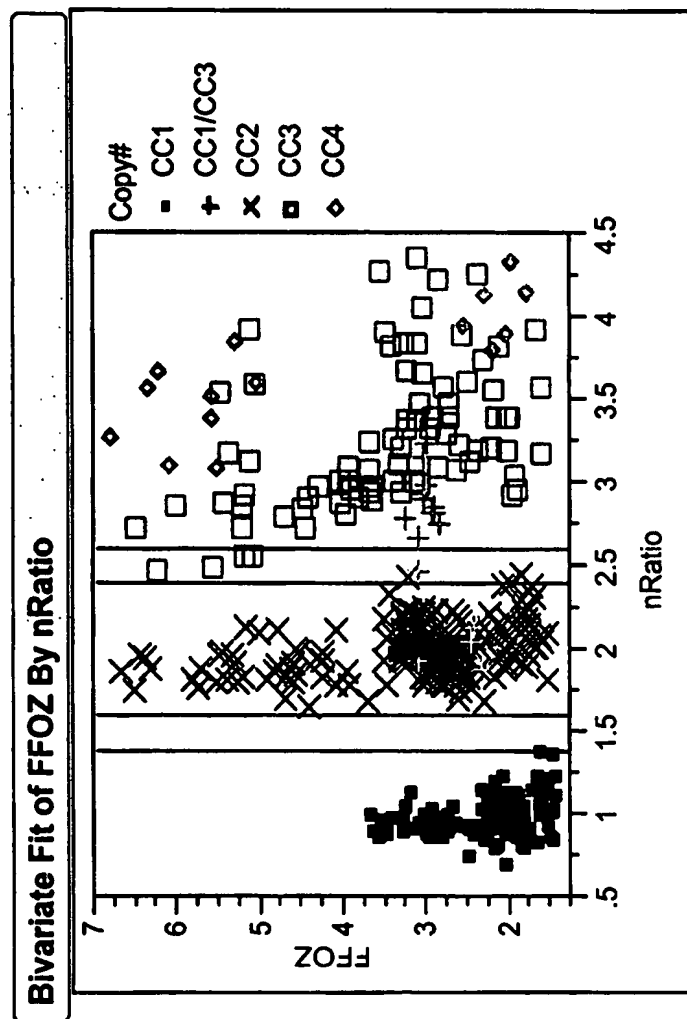
			Figure 12		SEQ ID NO:
Chrom	Gene	Accession #	Cytoband	Target Sequence	
1	alpha actin (ACTA1)	M20543	1q42.13	(ATGTCACAGGGTTCC)AACAGGCCACTCTTCCCTGGATGG)	203
1	HIST2H2B E	NM_003528	1q21.2	GTATCCACAGGA(GGCCCCCATCGCACGGCAGAACT)GJGTGACCGGACACA AAAGCTGCTGCCAAGCGTCAGTCATACAGAGCTGTACAGCAGT	204
13	DLEU1	NM_005887	13q14.2	G(GCACATGCCGCGAGAT)CATCGTGGTGACGGCTCTCCCT)TTGCTTCTT CGGTTGCAGTCTCTTCTTCTTGGCGTGGTGTAGCGCTTT	205
13	PCDH9	NM_020403	13q21.33	ATGGTAACGTTGCTTCGACGAGGAGTGJAGCTGCCGTCACTAGCCAGC ACTGT)CACCTTTGTGAATGGCTGTCTCTCTATCTAAGGACC	206
18	FLJ23403	NM_022068	18p11.22	GCCAACACTTCAGGATGAATATGTGAGCATAGATGTCTCT(CCACACAGATC CAGCTGGACAGGCTCAAAGTTGTCTCGTCCA)CACCCAGTC	207
X	PFKFB1	NM_002625	Xp11.21	ATGGTCTCTTCATGTGACTGGTCCACACCTTCAGGAGCTG)ATGCC TGGGACTGAATGAAGTTGGCCA)GGGCATAGGCA	208
Y	PRKY	NM_002760	Yp11.2	CTCCTTCT(GCACCCAGAGTAGGGGTGGAGTATACCCCTT)PCCATGTGT AGTGAATATG)GTGTCAGTGAGAGCCAGTTGGTCCCTCGGC	209
21	NRIP1	NM_003489	21q11.2	GGCTCCGATTAAAGTCTTCG(GACACTGGTAAGGCAG)GTGCGCTTCTC TGACACAGCAGGAGC)CATACCAAGAATGGGCACTCTTAGCAT	210
21	HLCS	NM_000411	21q22.13	GAGGATG(AGGTTTCTCAGCATGT)GAAAGGAGTTGCCGTCCGGGTGCA CAGTCACAACCTCGCCGCCCTCCTGGTGAACCTGGAGGAAGCCA	211
18	CN2	NM_018235	18q22.3	TGGCC(GGAGAAGAGAGGCGAAATCAGGAGGATGATGGAAG)TTGCTGC TGACAGAT)GTTAAGCAGTTGGGGGGCTCTGTGGAAGTGGTGATA	212
X	MTMR8	NM_017677	Xq11.2	CCTTGGGTACCGTAATATGAT(CCATGACTGCAGTTCCCGCCAC)CGAA GATCTCAGTCT)ACTCCAGATGCCCGCCACCGGTCTAGCCG	213
X	FLJ21174	NM_024863	Xq22.2	GAAGAGTTCAAGTCTGCTAGGTTCTTATTTT(GGACTCTCATGATGCT)GAG GTCAAGATGCCAGCTGGAGGCCA)GGCGCCGAGCTCACGCCCT	214
X	PCTK1	NM_033018	Xp11.3	GAGATTGTCACGAGGAGTGAAGATGGGGTCTGATGGGAGAGTGC CAGGCTTCA)GCCACGTCCTCGGATGAGGTGCAG)TCTCCAGTGA	215
Y	SRY	NM_003140	Yp11.31	TCATCCCTGTACAACTGTGTCCAGTGCACCTCGCT(GCAGAGTACCG AAG)CGGGATCTCGGGGAAGCAACTGCAATTC)TTC)SSCAGCA	216

Figure 13A
Chromosome Xp Invader Assay:PFKFB1+PCTK1 –Varying DNA Levels



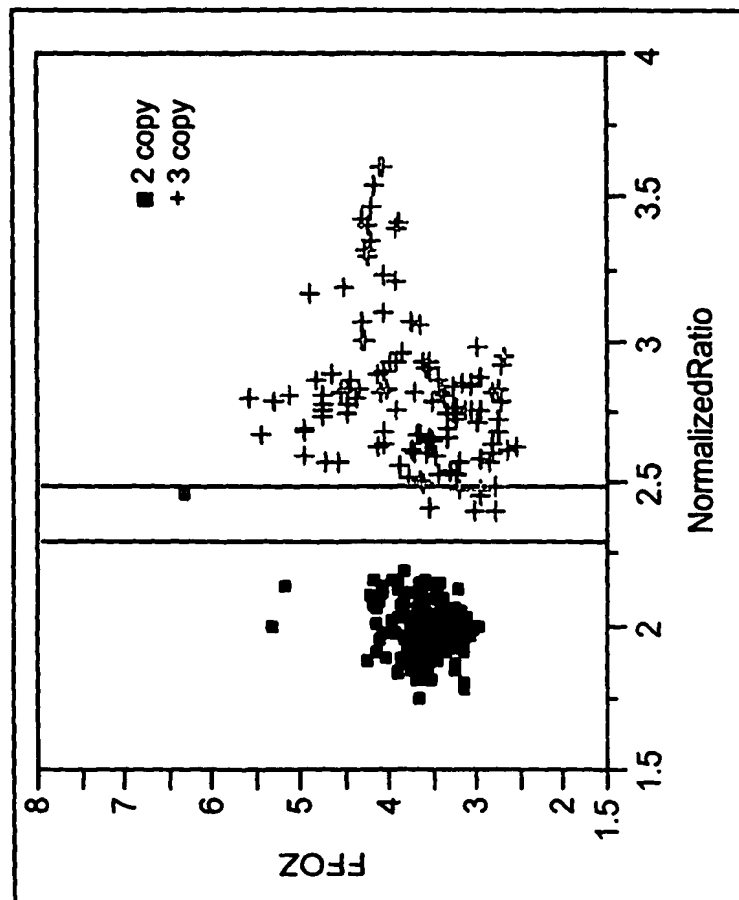
- 3 - 30 ng of DNA per reaction
- Equivocal zones: 1.4 - 1.6; 2.3 - 2.5
- N=637 samples, 517 normal, 120 aneuploid
- No call rate 1.9% (7 samples < 1.4 FOZ, 5 equivocal samples)
- Miscall rate 0%

Figure 13B
Chromosome Xq Invader Assay:MTMR8+FLJ21174 –Varying DNA Levels



- 3 - 30 ng of DNA per reaction
- Equivocal zones: 1.4 - 1.6; 2.4 - 2.6
- N=638 samples, 518 normal, 120 aneuploid
- No call rate 3.3% (15 samples < 1.4 FOZ, 6 equivocal samples)
- Miscall rate 0%

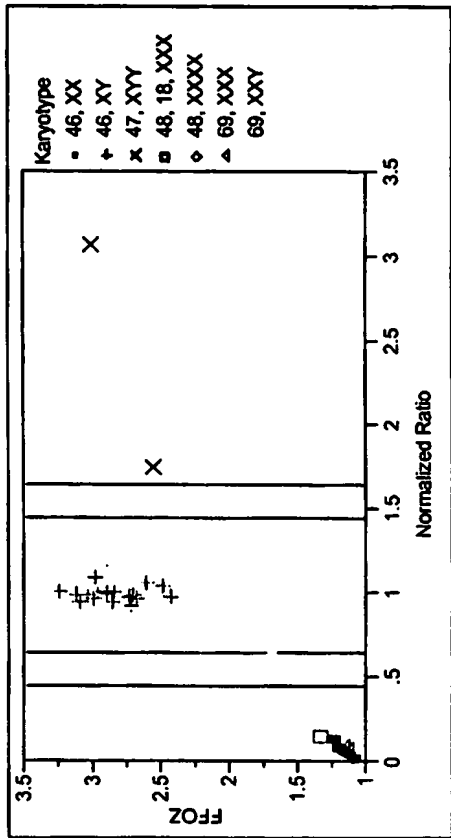
Figure 14. Chromosome 18 Invader® Assay – Sample Mixtures



- Trisomy 18 / Disomy sample mixtures
- 10 ng of DNA per reaction
- Equivocal zone: 2.3 – 2.5
- N=315 samples, 198 disomy, 117 trisomy (+18 with 0%, 10% and 20% disomy contamination)
- No call rate 2.9%
- Miscall rate 0%

Figure 15: Analysis of Triploidy Samples (69, XXY)

Normalized Ratio Analysis

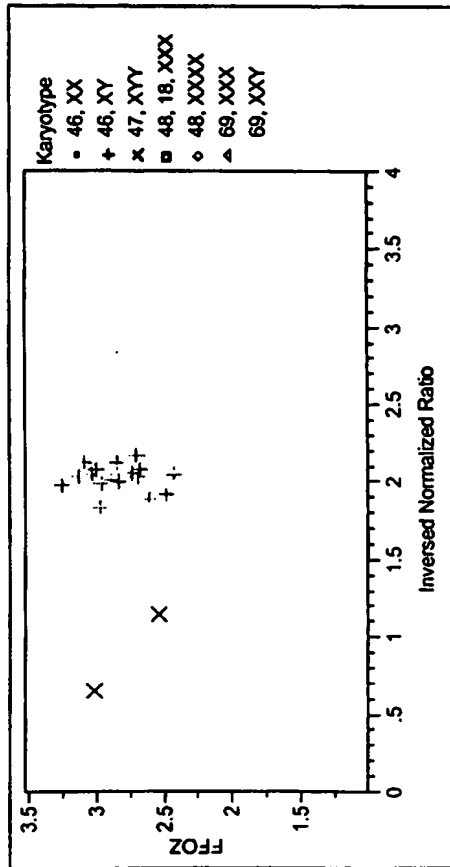


$$\text{Ratio} = \frac{(\text{Net chromosome specific FOZ})}{(\text{Net internal control FOZ})}$$

where Net FOZ = FOZ - 1

$$\text{Normalized Ratio} = \frac{(\text{Ratio of unknown sample})}{(\text{Ratio of male control sample})} \quad \text{X (1)}$$

Inversed Normalized Ratio Analysis



$$\text{Inversed Ratio} = \frac{(\text{Net internal control FOZ})}{(\text{Net chromosome specific FOZ})}$$

where Net FOZ = FOZ - 1

Inversed Normalized Ratio =

$$\frac{(\text{Ratio of unknown sample})}{(\text{Ratio of male control sample})} \quad \text{X (2)}$$

Note: Samples not containing a Y chr. not shown on graph due to high inversed normalized ratios (>10)